Molecular Biology

THE 22q11.2 DELETION SYNDROME AND SCHIZOPHRENIA

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The 22q11.2 Deletion Syndrome, including velo-cardio facial syndrome (VCFS), is the most common chromosomal deletion syndrome in humans. A deletion of this region induces specific phenotypic changes including a cleft palate, limb and digit anomalies, and cardiac defects. Individuals with VCFS have an increased incidence of schizophrenia than the general population. This increased incidence suggests an enhanced vulnerability to schizophrenia among individuals with VCFS, which may reveal a genetic basis for schizophrenia.

There is a syntenic region to the 22q11 deletion region on murine chromosome 16. A mouse model was used in an effort to understand the potential role of each gene in schizophrenia by examining mice hemizygous for the chromosome 16 region. The etiology of schizophrenia in humans is poorly understood, but there are several hypotheses that may shed light on this disorder. The cerebrovasculature hypothesis theorizes that due to the cardiac defects in 22q11 deletion syndrome patients, there should be an increase in the amount of cerebrovasculature formation. The decreased amount of oxygen to the brain should result in the increased amount of blood vessels in mutant mice. To test this theory, E10.5 embryos were subjected to the whole mount embryonic staining protocol using the PECAM-1 (platelet endothelial cell adhesion molecule) antibody. The structure of the cerebrovasculature was examined quantitatively. The anastomoses of major blood vessels in the brain were counted to compare the structural formation of the cerebrovasculature in both wild-type and mutant mice. In conclusion, the mutant mice for the 22q11.2 Deletion Syndrome displayed a higher amount of cerebrovasculature formation than did their wild-type counterparts.